

GENETICS AND NEWBORN SCREENING ADVISORY COUNCIL MEETING

The Genetics and Newborn Screening Advisory Council meeting was held on Friday, February 28th, 2014 at the Florida Department of Health Bureau of Laboratories, 1217 Pearl St., Jacksonville, FL.

Call to Order:

The meeting was called to order at 10:07am EST by Paul Pitel, MD, Chairman of the Council. Roll was taken and introductions were made.

Members Present:

Paul Pitel, MD, Chairman, Jacksonville
John Waidner, MD, Jacksonville
Robert Fifer, PhD, Miami (UM)
David Auerbach, MD, Orlando
Dorothy Shulman, MD, St. Petersburg (USF)
Keith Nash, Orlando, March of Dimes
Bonnie Hudak, MD, Jacksonville
Susanne Crowe, MHA, Jacksonville
Roberto Zori, MD, University of Florida
Melissa Perez, Tallahassee
Lori Kohler, RN, representing Lori Gephart, RN, Agency for Persons with Disabilities
Celeste Philip, MD, MPH, Deputy Secretary for Health, Tallahassee (via teleconference)
Heather Smith, Lakeland (via teleconference)

Guests:

Linda Carter, PerkinElmer, Inc.
Jeanne Brunger, PerkinElmer, Inc.
Bianca Gomez, Audiology Extern, University of Miami
Cory McNabb, Audiology Extern, University of Miami
Sarah Guillemette, Audiology Extern, University of Miami
Audrey Hall, Clarke Schools for Hearing and Speech, Jacksonville
Cynthia Robinson,
Fiona Mackay, March of Dimes
Larry Vroegindewey, DOH SCID
Lucy Raub, University of Florida
Sharon Bowden, Pediatrix Medical Group
Susan Weinger, Pediatrix Medical Group

DOH Personnel Present:

Lois Taylor, RN, CMS, Tallahassee
Whitney G. Jones, CMS, Tallahassee
Drew Richardson, CMS, Tallahassee

Jasmin Torres, Bureau of Laboratories, Jacksonville
Ming Chan, PhD, Bureau of Laboratories, Jacksonville
Jojo Dy, MD, Bureau of Laboratories, Jacksonville
Patty Parrish, Bureau of Laboratories, Jacksonville
Chrishonda Jenkins, RN, CMS, Tallahassee (via teleconference)

Housekeeping/Reminders

Dr. Pitel reminded council members and guests to hand in their money for lunch and to review the minutes of the July 2013 council meeting for approval before the break.

Introductions:

Dr. Pitel introduced Celeste Philip, MD, Deputy Secretary of Health and Deputy State Health Officer for CMS, as the new CMS Representative for the Genetics and Newborn Screening Advisory Council.

2014 Legislative Update

Dr. Philip provided an update on the recent legislative activities in regards to the Department of Health and Children's Medical Services. Proposed changes to 383.14 are to change the word "physician" to "provider" to correct the limitation of who can receive newborn screening results electronically and via other media such as audiologists, midwives, etc. Changes were also proposed for 383.145 for the Newborn Hearing Screening Program to update old language and update the range for hearing loss. These changes are proposed to be consistent with national standards.

Dr. Philip made a presentation to the Health Quality Policy committee. The committee was very engaged and was very appreciative of the value of the program and the efforts made to care for Florida's newborns.

In the Governor's Budget for CMS there is a 2.8 million dollar for Child Protection Teams (CPT) to increase the number of staff to help with assessments and quality assurance and improvement. There is also a 2 million dollar request for budget authority allows the Department of Health to use money for the Healthiest Weight initiative from grants or donations that may be received in the coming year. The Healthiest weight initiative is a public/private partnership that helps create environments that make it easier for people to make healthier choices. 3 Million dollars has also been appropriated for Alzheimer's research.

A question was asked if anything was being done for genetic counselors in the legislature. Dr. Pitel stated that it is not a funding issue but issues with licensure for

genetic counselors. Since genetic counselors are not licensed they cannot bill insurance for services.

It was mentioned that there were ongoing issues with funding for the genetic centers and the future of the centers.

CCHD Implementation Update

Dr. Pitel provided an update regarding the CCHD implementation. The council gained support from State Surgeon General, Dr. Armstrong, for adding CCHD to the list of disorders screened in Florida. The council was tasked with putting together a joint workgroup that met several times via conference call. The workgroup created a good plan for Florida based on current literature. CCHD screening is very much like hearing screening in that the screening is performed in the hospitals. With CCHD, there must be immediate intervention if the newborn fails the pulse oximetry screening.

The screening test will not detect every newborn with CCHD but it will detect many types of CCHD. 100 of the 119 hospitals have already implemented a CCHD screening procedure with a one or two site testing. The target groups for this screening are babies in the well baby nursery that otherwise look healthy, but have an underlying issue not visibly present or show signs or symptoms. The recommended screening is to test the right hand then either the right or left foot. The algorithm recommended by the American Academy of Pediatrics provides information to determine if the results are normal or when the baby needs to be referred for further evaluation. The funding approved by the 2013 Legislature for CCHD is to support data collection and surveillance.

The workgroup recommended that Florida should adopt the national standards that specify the two site testing algorithm. There was discussion among the group regarding how to test babies in the NICU and what the term “timely referral” meant in the recommended documents. The suggested language left the decision up to the treating physicians to make the referral based on their clinical judgment of each individual situation. The recommendation specifies that the CCHD screening should be performed when the newborn is over 24 hours of age. However discharge times are increasingly rolling back to 24 hours so it increases the chance that the baby may have the screening prior to 24 hours or not at all. It is the responsibility of the providers to ensure that this test is performed in a timely manner. All newborn screening tests are mandatory unless the parent objects in writing.

SCID Update

The Newborn Screening Program began screening for SCID October 2012. The protocol to classify results as low or inconclusive was combined into one category. Also, if a baby has three specimen results with inconclusive or borderline results then

they are referred to the referral center. Over the last 15 months the program noticed that one out of every three referrals came from one specific hospital. It was discovered that the hospital was drawing blood from an arterial line that contained heparin. This resulted in abnormal results. Education was provided to that facility and there have been a drop in the number of abnormal SCID borderline and presumptive positive results generating from that facility. Currently Florida is right on target for the number of babies being diagnosed which is 2-4 per year. For presumptive positives if the baby has a previous normal or a repeat normal screening then the case is closed.

The council discussed how the program followed up on presumptive positive results and referred them directly to the referral centers and how these results are given in a timely manner. Primary care physicians have access to the results. The Newborn Screening Program sent out 15,000 letters and emails to all physicians in the state to inform them of SCID being added to the screening panel. Borderline result patients get a phone call from a Newborn Screening Follow-Up Nurse after the results are released by the laboratory. For presumptive positive results the follow-up nurses call the SCID referral center after the results are released. There are monthly conference calls with the SCID centers to discuss cases.

Cystic Fibrosis Update

Dr. Hudak gave an update regarding Cystic Fibrosis. A year ago a review was conducted of the Cystic Fibrosis mutations found in Florida. The review found that Florida was good with detecting CF in newborns of Caucasian background, but not as well at detecting it in newborns of African American descent. There was one mutation that occurred a few times among black infants, the A559T mutation. European literature has reported that this mutation happens more often among infants of African descent. The Cystic Fibrosis contract is up for renewal in 2015. Dr. Hudak made a motion that mutation A559T be added to the CF mutation panel. Dr. Fifer seconded the motion to add a new mutation. Vote was unanimous.

Newborn Screening Laboratory Update

The lab resumed Saturday testing on January 11, 2014. Friday specimen results are reviewed and released on Saturday and results for specimens received on Saturday are released on Monday. Since October 2012 when Florida started screening for SCID there have been one confirmed x-linked SCID, one immunodeficiency, two variants, and four Chyllothorax, with 256 early/inconclusive cases, and 29 borderline cases.

In November of 2013 the lab began reporting Total Galactose for the purpose of reducing the number of referrals made. Ms. Taylor reported some confusion from the referral centers regarding that the total galactose numbers. It was brought to the council's attention that one of the referral centers received a borderline case with negative total galactose which caused confusion. The lab agreed to look into that case.

On January 30, 2014 testing for Biotinidase was transferred to an automated analyzer. The benefit of this is reducing the number of ways of processing so more specimens are moved into each space and the processing time is reduced. There is also less human error since there are fewer hands on the card. This coming March a demonstrator will be here to demonstrate a possible second-tier method of testing. In 2013 PerkinElmer offered a workflow assessment of the lab. Their recommendation was to provide more extensive trainings and maximizing the use of the punchers. Right now the lab is splitting the specimens between the 9 plate punchers and the three plate punchers. The lab has already implemented these recommendations and the lab has had refresher trainings from the manufacturers for the use of the instruments. Other recommendations were to continue follow up on unsatisfactory specimens and to work on more efficient ways for hospitals to send specimens to the lab. The lab does not have a contract with a courier business and it is up to the hospital to do this. Some couriers send the specimens to Memphis first before delivering them to the lab and the lab is working to see if they can be the first drop off point not the second.

The lab is participating in the LEAN project which is a 20 piece approach that started in 2014. Right now the lab is using four keys which are organizing, error proofing, time management, and SMART Expectations.

Newborn Screening Follow-Up Program Update

NewSTEPs received a grant from HRSA to provide technical assistance to newborn screening programs. Florida was the third state to be reviewed. Overall they were impressed with our program. They were impressed with the referral centers, the funding of the program, and thought the Advisory Council increased the value of the program. Florida is the only state that bills Medicaid and private insurance for testing. NewSTEPs was impressed with Winnie Palmer hospital and their education efforts. They mentioned that the lab staff is dedicated and flexible and said very good things about the turnaround time for testing. They suggested that the program review specimen storage and use policy and they would like to see the unsatisfactory rate below 1%. A formal report will be ready in about four months.

The unsatisfactory rate has increased. There has also been an increase in the number of referrals to the referral centers. The program has stopped following DOB Hypothyroidism because most are in the NICU and will have a repeat specimen before hospital discharge. So far there have been more 25,000 searches done in FNSR and about 7,000 are done in the office.

Although Pompe Disease was recommended by the Secretary's Advisory Committee on Heritable Disorders for Newborns and Children on May 17, 2013, it was not added to the Recommended Uniform Screening Panel by US Department of Health and Human Services Secretary Kathleen Sebelius. She moved the issue to an internal committee

for review and approval. It was reported that there was not a unanimous vote when the SACHDNC recommended it.

Staffing issues for the Follow-Up program is no longer a problem as the program no longer has vacancies. However the staffing contract was lost and five positions were moved over to OPS positions.

Ms. Taylor also gave an update for the Newborn Hearing Screening Program. The Early Hearing Detection and Intervention (EHDI) national meeting will be held in Jacksonville in April. Many states receive federal grants to fund their program and Pam Tempson is the coordinator of the EHDI program in Florida. Hearing screening results can be reported using the eReports website and more people are beginning to use it around the state.

The Department of Health celebrated its 125th year of being in the public health business. Newborn Screening will celebrate its 50th year in 2015. Newborn Screening began in Florida in 1965 when the Lab began PKU testing.

eReports, DSM, ELO/ELR Update

Drew Richardson provided an update on the Newborn Screening systems. Florida Newborn Screening Results (FNSR) website continues to work well and is responsible for 95% of the requested results from physicians. eReports is used to report hearing screening results and went into production June 2013. Of the 442 users about 35% are trained in the system. With the 35% trained, the program is receiving greater than 50% of the repeat hearing screening results because the high volume submitters were targeted for training. Training is provided by Laura Olsen, Hospital Hearing Educator. eReports will also be modified to include the submission of pulse oximetry screening results that are not submitted on the specimen cards. The projected go-live date for collecting the pulse oximetry results is June 2014.

Electronic Laboratory Ordering/ Electronic Laboratory Reporting (ELO/ELR) project has received live data for testing. There are still areas that need to be completed in this project. Winnie Palmer Hospital has indicated interest to use ELO/ELR.

The APHL community created a Health Information Technology workgroup. Drew Richardson is a member and serves as a co-chair for the workgroup. The workgroup collaborates with all states and many vendors to pool knowledge together for better use of technology for newborn screening.

Screening Center will be a large upgrade to the lab's database and a minor upgrade for the Follow-up Program. Screening Center will allow us to create our own reports. Direct Secure Messaging (DSM) is a program that allows us to send and receive secure messages from our referral centers rather than having to fax these referrals. DMS allows the follow-up program better tracking on referrals.

New Discussion Items

The advisory council reviewed the minutes from the July 2013 meeting. The minutes were approved with minor changes to be made.

Dr. Zori proposed that a biochemical geneticist be available for consultation to the lab in a formal role. Lois commented that this has been recommended several times by those who reviewed the program and there have been instances when a geneticist was working with the lab on a part time basis. Dr. Pitel motioned for the lab to craft a proposal to explore hiring a biochemical geneticist. Dr. Hudak seconded the motion. Vote was unanimous.

Dr. Pitel spoke to the council about the possibility of getting funding for a Fragile X Project study. The council discussed the requirements for passing a study through DOH for approval and possibilities for the newborn screening program to do this study.

Public Comments/closing/summary

Cynthia Robinson of Clarke Schools in Jacksonville gave a presentation about their schools to the council. Dr. Pitel thanked them for the information and their presentation.

Adjournment

The meeting adjourned at 2:13pm EST.